

Am. J. Hum. Genet. 63:1920–1920, 1998

BOOK REVIEWS

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Genetic Disorders and Pregnancy Outcome. Edited by L.D. Platt, R. Koch, and F. de la Cruz. New York: Parthenon Publishing Group, 1997. Pp. 146. \$85.00.

This slim, well-written, and nicely illustrated book represents a gold mine for anyone interested in metabolic diseases and pregnancy outcome. However, the interested person may find it hard to pick up this book, because of its somewhat misleading and nonspecific title. Several aspects of pregnancy in patients with five different metabolic diseases, as well as the effect that molecular-genetic screening and successful postnatal treatment of phenylketonuria (PKU) homozygotes have on the gene pool, are reviewed in this book. The book focuses mostly on pregnancy and PKU, ovarian insufficiency associated with galactosemia, pregnancy in Gaucher disease, and diabetes mellitus. An excellent historical overview of both fetal and maternal metabolic diseases in pregnancy is given in the first chapter. The following eight chapters, dedicated to PKU, summarize the most recent data from the Maternal Phenylketonuria Collaborative Study, whereas other chapters discuss reproductive issues in ornithine transcarbamylase deficiency, diabetes mellitus, galactosemia, and Gaucher disease. Although many different obstetric issues of maternal PKU are presented, the book provides only limited information about practical issues of

daily patient management, such as dietary prescription, use of supplements, frequency of follow-up, and so on. The chapter on ornithine transcarbamylase deficiency focuses on the maternal phenotype, rather than on pregnancy and its outcome. The only criticism that I would consider is that a single chapter on embryopathy in women with diabetes seems out of place in this short volume, because the breadth of available data in this area easily merits an entire textbook of its own. This book can be highly recommended both as an introduction to metabolic disease in pregnancy, for residents and fellows in obstetrics and family practice, and as a reference for geneticists, endocrinologists, and obstetricians who may be facing the challenge of managing the pregnancies of patients with these metabolic diseases.

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